Current Oversight of Genetic Testing

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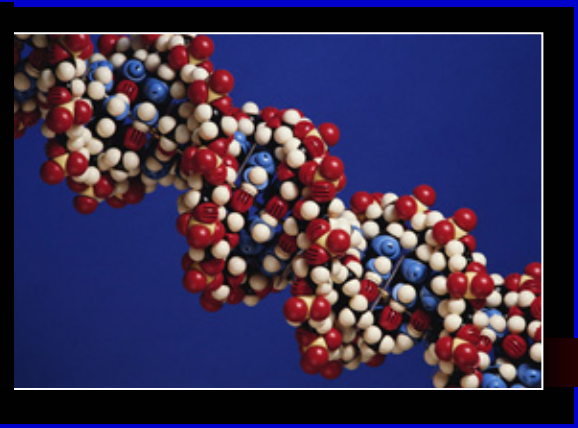
GALLEGO



Genetic Testing Oversight Issues

- Genetic revolution – New scientific, medical, social, legal, ethical concerns
- Genetic evolution – potential benefits and potential risks

Challenge:

Bring our public policies in line with the genetic revolution

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Quality Testing

Access

Cost

Positive aspects of regulation/oversight

- Protection of the public – Sanction
- Level playing field – Minimum Standards
- Provide benchmarks for good practice
- Monitor attainment of goals (PT, QA, QC)

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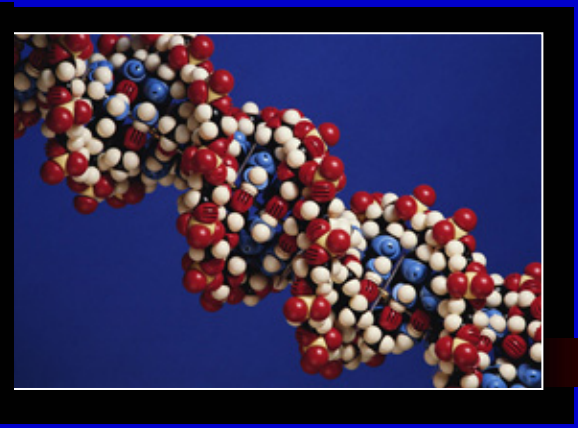

Negative aspects of regulation/oversight

- Always out of date
- Focus on process rather than outcome
- Increases costs
- May not prevent bad outcomes
- May impede new technology
- May impose rigid requirements (personnel)

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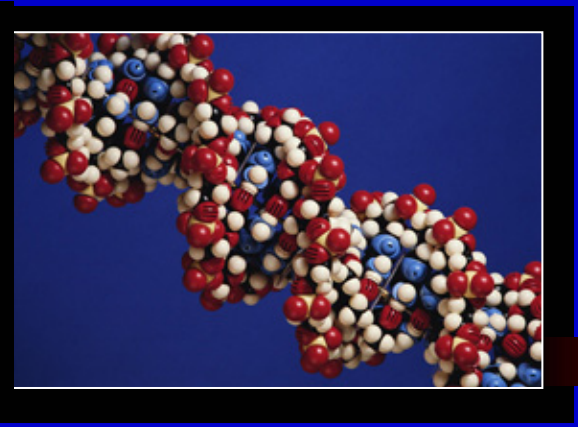

Background

- CLIA enacted - 1988
- NIH/DOE Task Force Report - 1997
- CLIAC recommends changes to CLIA – 1998
- SACGT recommends increased oversight – 1999
- CDC Notice of Intent – May 2000

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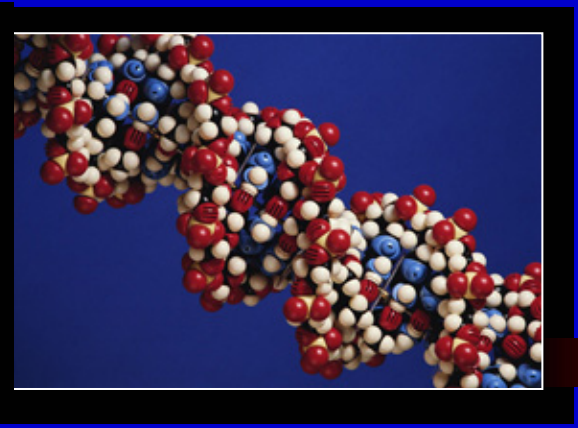

NIH/DOE Report: Areas of Concern

- Appropriate introduction of new genetic tests into clinical practice
- Adequate regulation of laboratory testing
- Increasing healthcare provider and patient understanding of genetics
- Maintaining access to quality testing for rare diseases

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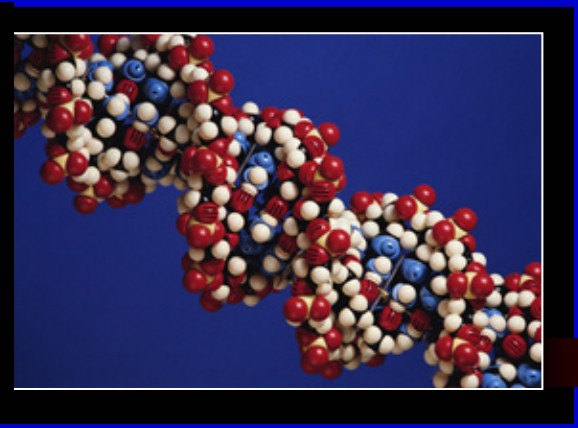

Proposed CLIA Genetics Specialty

- Definition – What is included and excluded?
- General requirements
  - Documentation of clinical validity
  - Person authorized to order a genetic test
  - Informed consent
  - Confidentiality
  - Genetic counseling
- Requirements for specific testing phases
  - Pre-analytic phase
  - Analytic phase
  - Post-analytic phase

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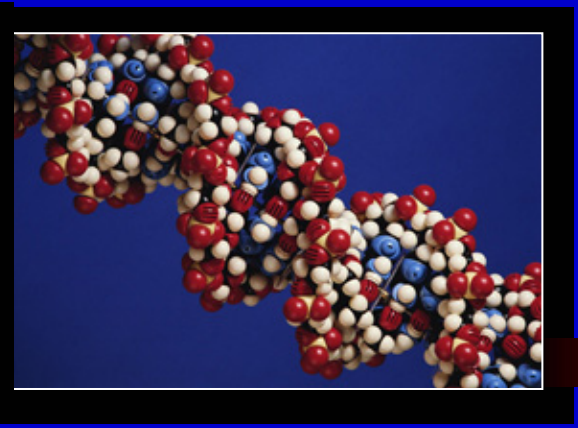

CLINICAL VALIDITY

1. Lab director's role in documenting clinical validity:

- **no responsibility for documentation; or**
- **ensures that documentation exists in literature;**

or

- **documents clinical validity of all tests offered**

2. Should clinical validity be established before a test can be offered?

- **mixed opinions on this issue**

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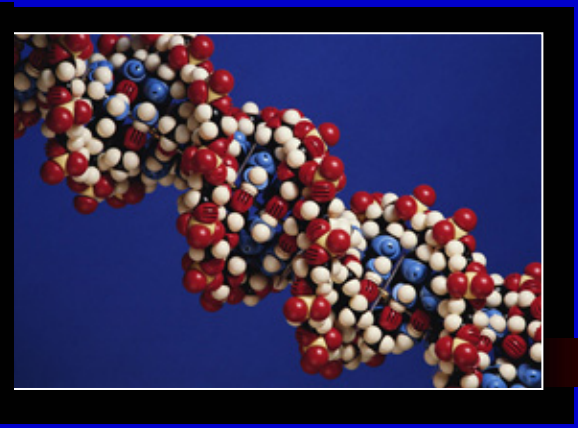

INFORMED CONSENT

1. **Should CLIA require documentation?**
   - **Is documentation an integral part of laboratory practice?**
   - **Is CLIA the place to "police" ordering physicians?**

2. **Should the laboratory's role in assuring documentation of IC include:**
   - **documentation that an authorized person has obtained IC?**
   - **alerting health care providers when IC is needed?**
   - **providing IC forms to health care providers?**
   - **documenting the adequacy of IC forms?**

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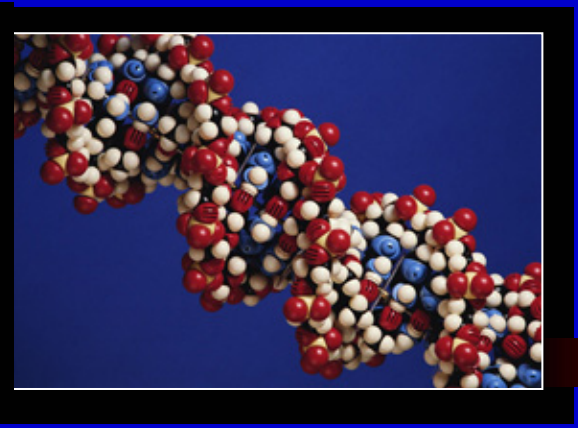

CLIAC Issues

- QA/QC/PT
- Re-use of samples
- Authorized person to order genetic tests
- Confidentiality
- Test requisition and clinical information
- Result reporting
- Record and specimen retention

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Secretary's Advisory Committee on Genetic Testing - Recommendations

- Strengthen human protection in research – IRB review and informed consent
- Augment CLIA to address genetic testing
- Establish FDA review of all new genetic tests
- Develop information on the clinical utility of genetic tests

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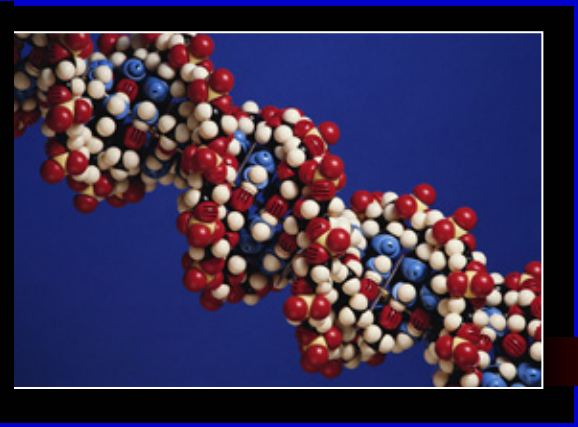

Current US Oversight of genetic testing

- CLIA for laboratories
- FDA for kits and devices
- IRB for patients in research
- NYS – QC, personnel, test validation, test review and approval
- Professional guidelines and standards of practice (AMP, ACMG, CAP, etc.)

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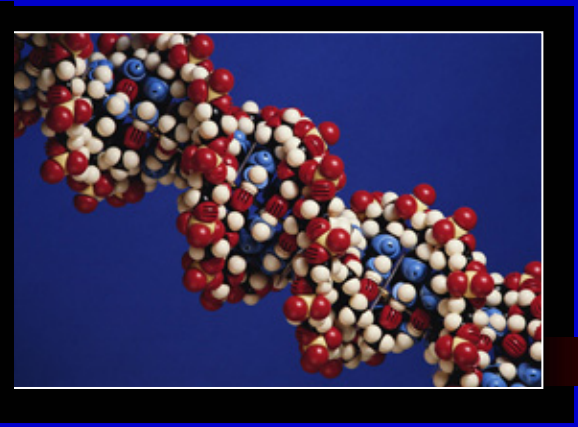

International oversight of genetic testing

- UNESCO
- OECD
- European Commission
- ISO
- ILAC/WHO
- Eurogenetest

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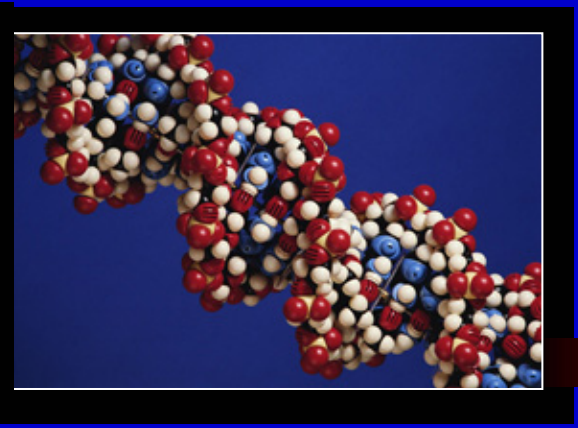

International oversight of genetic testing

- WHO
- Professional guidelines and standards of practice
- Sweden/Norway Biobanks
- Others

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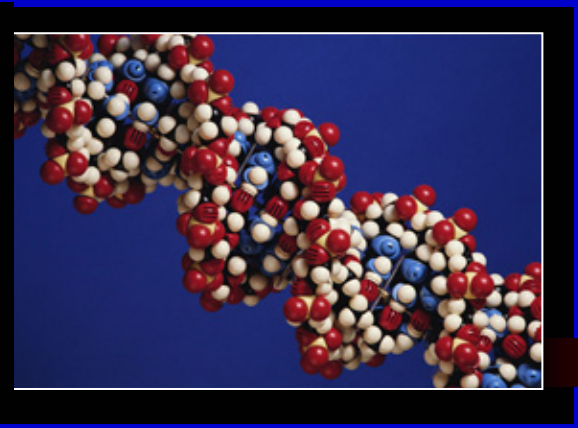

Secretary's Advisory Committee on Genetics, Health and Society – Additional Concerns

- Coverage and Reimbursement
- Large Population Studies
- Pharmacogenomics
- Direct-to-Consumer Marketing

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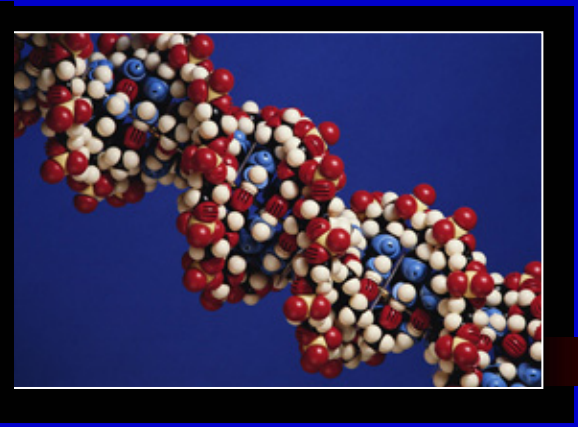

The Genetic Testing Environment

- Rapid advances in genetic technology
- Molecular basis of both rare and common disorders
- Commercialization of testing
- Genetic testing no longer for rare diseases or conditions

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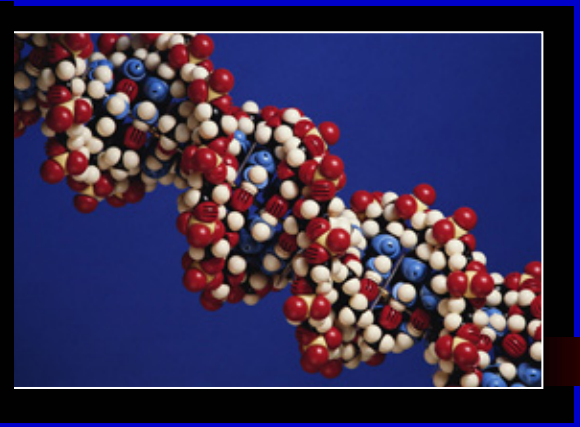

Public Policy Challenges

- Balancing access, costs, and quality of services
- Clarifying roles of government, professional organizations, advocacy groups in ensuring adequate oversight
- Dealing with new issues posed by genetic testing
- Obtaining data needed to guide policy decisions

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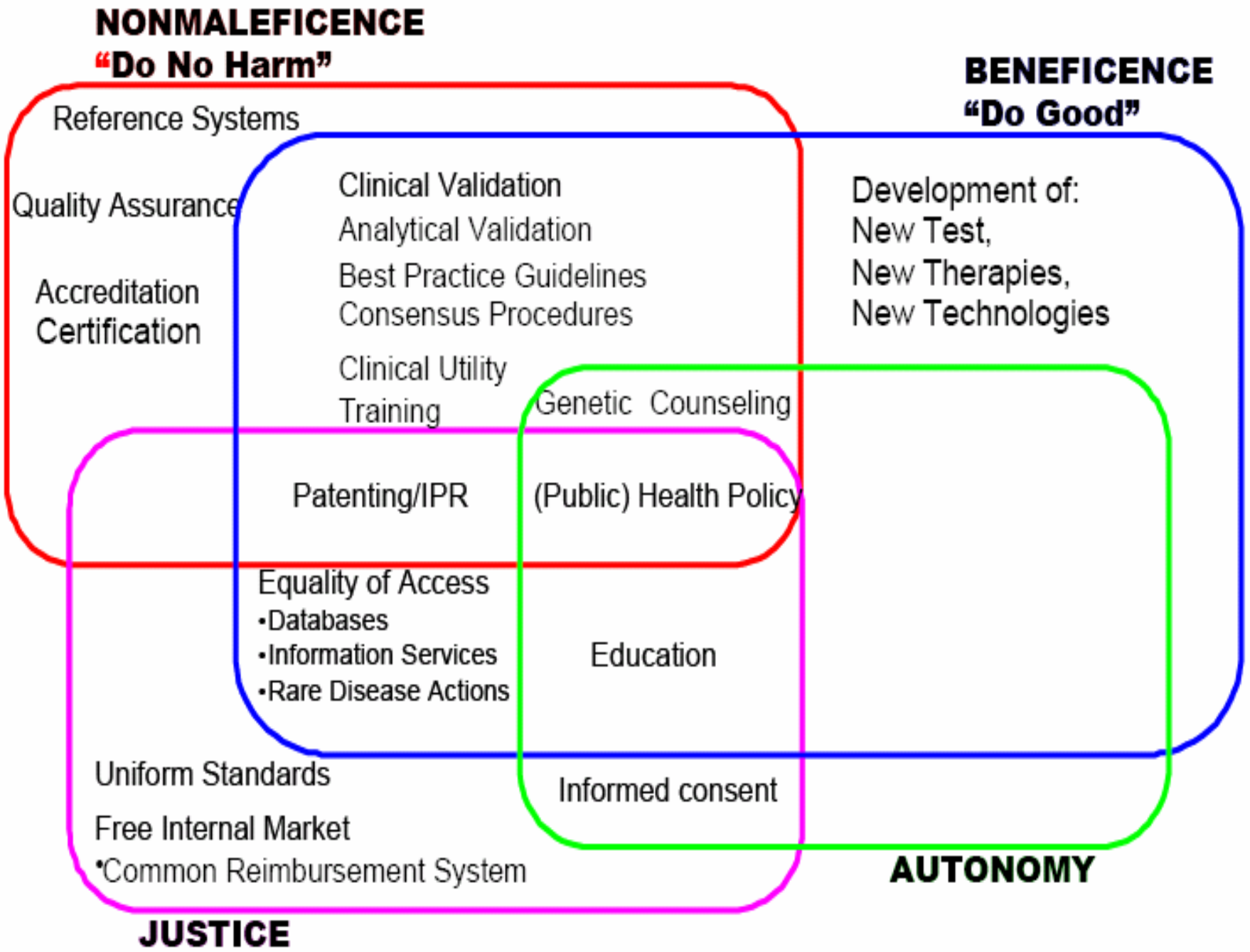
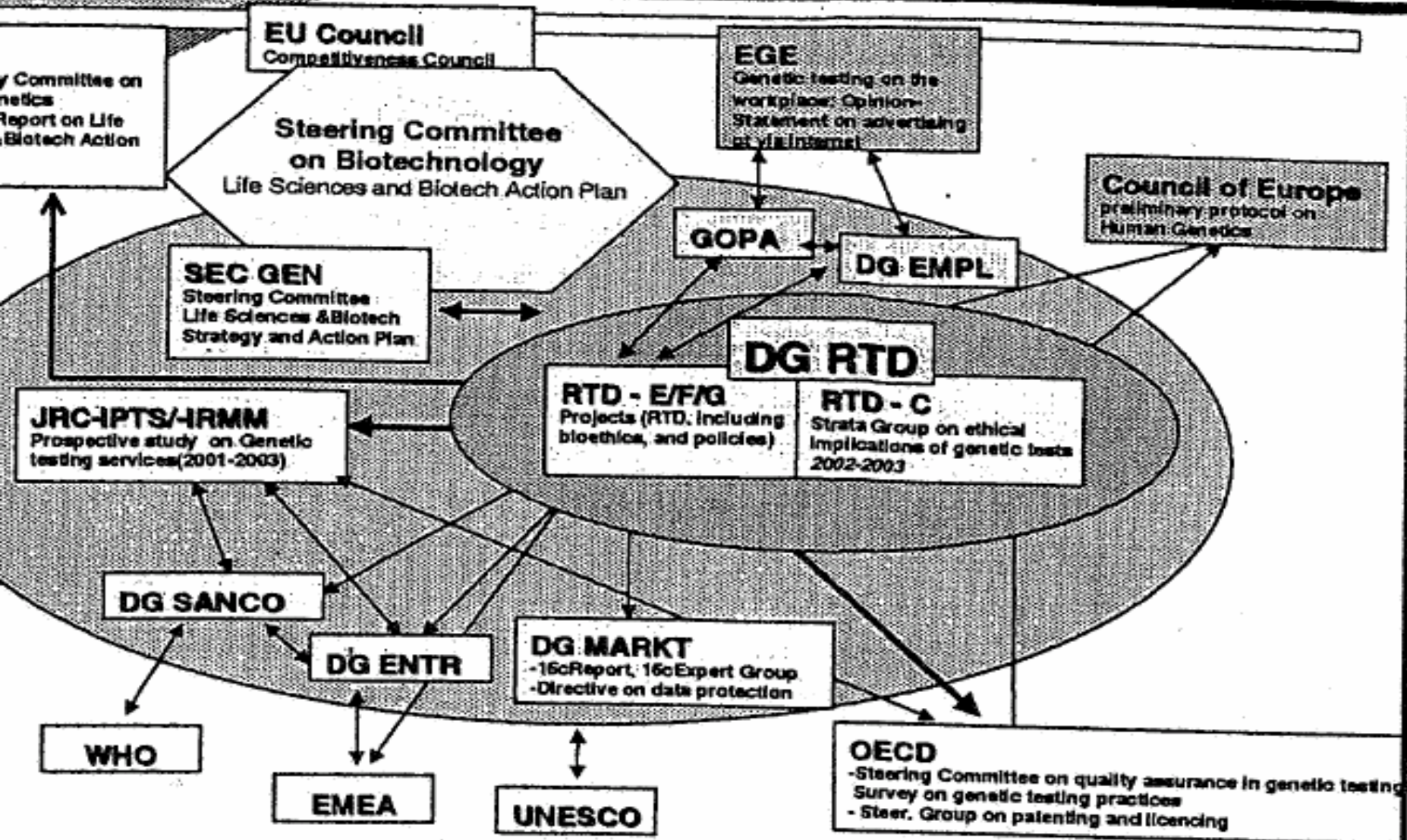

NONMALEFICENCE
"Do No Harm"
BENEFICENCE
"Do Good"
Reference Systems
Quality Assurance
Accreditation
Certification
Clinical Validation
Analytical Validation
Best Practice Guidelines
Consensus Procedures
Clinical Utility
Training
Development of:
New Test,
New Therapies,
New Technologies
Genetic Counseling
Patenting/IPR
(Public) Health Policy
Equality of Access
•Databases
•Information Services
•Rare Disease Actions
Education
Uniform Standards
Free Internal Market
•Common Reimbursement System
Informed consent
AUTONOMY
JUSTICE

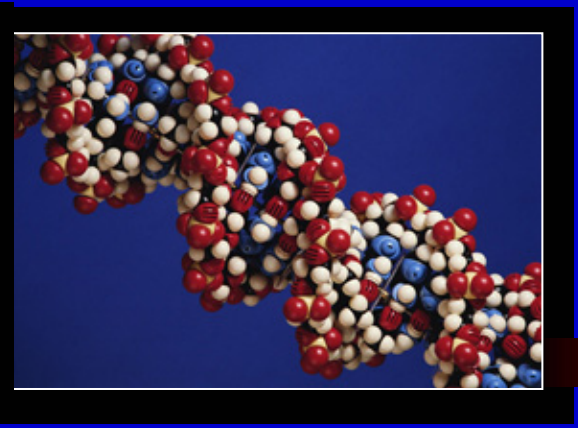
EUROPEAN COMMISSION

Genetic Testing Quality Assurance

Network of interactions



ernational organisations, institutions and EC services involved in genetic testing quality assurance in the EU



Observations

- Public/private partnerships are going to be important to the oversight process of genetic tests and testing
- Government oversight of labs under CLIA, tests under FDA, and human subjects under IRB will be enhanced
- Data will be needed to guide public policy decisions about when tests should be used to test individuals and populations
- ELS Issues will challenge society

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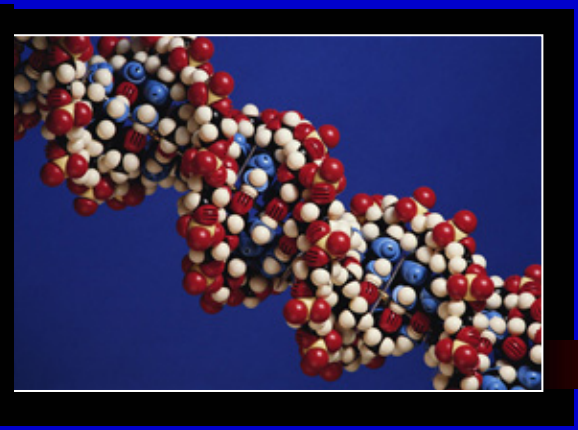

Oversight Goals

- Balance: Access = Burden/Protection
- Timeliness: With Broad Input
- Long-lasting: Crystal Ball
- Effective Implementation

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